

# Seminoma in a Male Phenotype 46XX True Hermaphrodite

Vinod Malik, Dinesh Gupta, Meenu Gill<sup>1</sup> and Amba Lal Salvi, Departments of Surgery and <sup>1</sup>Pathology, PGIMS, Rohtak (Haryana), India.

Although true hermaphrodite is uncommon, it has been reported in more than 400 individuals. Tumours arising in the gonads of true hermaphrodite is a very rare finding and only very few cases have been reported in the literature. We report a case of a 35-year-old phenotype man with bilateral undescended testis with lump hypogastrium. On examination, he had an ovotestis on one side and pure seminoma arising in testis on the other side. The seminoma arising in the left undescended testis was successfully treated by excision and combination chemotherapy. Cytogenetic studies revealed that 46XX karyotype with primer specific for the sex-determining region of the Y chromosome was negative. The karyotypic abnormality noticed in the patient is also reviewed in the article. [*Asian J Surg* 2007;30(1):85-7]

**Key Words:** ovotestis, true hermaphrodite, seminoma testis

## Introduction

Although true hermaphrodite is uncommon, it has been reported in more than 400 individuals. Tumour arising in the gonads of a true hermaphrodite is a very rare finding and only a few cases have been reported in the literature.

## Case report

A 35-year-old phenotype man presented with complaints of lump hypogastrium for 1 month with mild pain in the lower abdomen for 15 days. Physical examination revealed a firm mass in the lower abdomen that was slightly mobile. No other lump was palpable in the rest of the abdomen. Chest X-ray was normal with no evidence of any metastasis. His height was 5 feet 10 inches and he weighed 130 pounds. Pubic hair was of male type. The penis was well developed and the urethra was in the normal position. The testes were absent in the scrotum since birth, nor were they found in the inguinal region. His scrotum was poorly developed.

The patient had been married for 15 years and had no children. According to the patient, he had normal erection and ejaculation. He had normal secondary sexual characteristics with no gynaecomastia. He had four brothers and one sister and all of them have children. He took no treatment for infertility.

Provisional diagnosis of tumour in undescended testis was made. Ultrasound revealed a mass in the left lower abdomen with ill-defined hyperechoic and hypoechoic areas without any lymph node metastasis. Tumour markers, mainly AFP, B-HCG, CEA and serum testosterone, were all normal. Semen examination revealed azoospermia.

Suspecting a malignant neoplasm involving the undescended testis, an exploratory laparotomy was performed, which revealed a large tumour 25 × 15 cm on the left side of the abdomen, which was excised. On further examination, the patient was found to have a uterus and a normal ovary (gross appearance) on the right side (Figure 1). Hysterectomy and salpingo-oophorectomy were performed. The cervix was rudimentary with no external opening in the perineum.

Address correspondence and reprint requests to Dr Vinod Malik, H. No-155, Sector-14, Rohtak (Haryana) 124001, India.

E-mail: drrvmalik@yahoo.co.in • Date of acceptance: 26 September 2005

Other testis could not be located and the rest of the abdomen was normal. The mass (specimen A) and uterus with ovary (specimen B) were sent for histopathological examination. No evidence of metastasis was found. Post-operative recovery was unremarkable. The patient received three cycles of chemotherapy (BEP regimen) without any evidence of metastasis at 18-month follow-up.

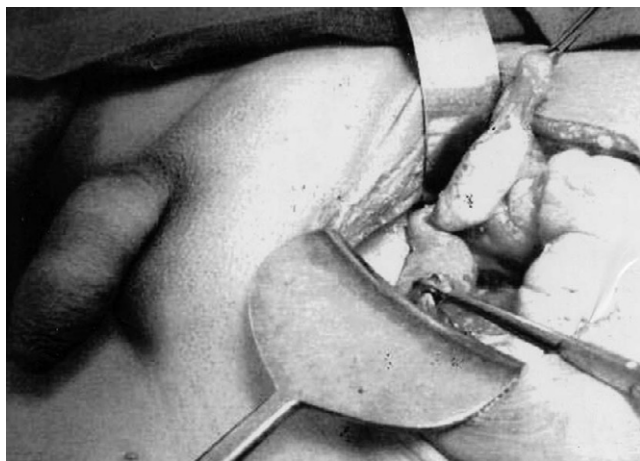
Pathological examination of the mass (specimen A) confirmed the diagnosis of pure seminoma extending through the tunica albuginea with stromal lymphocytic infiltration. Specimen B, which included the uterus and right tubo-ovarian nodule, revealed endometrium in proliferative phase, myometrium without pathology, cervix with mild nonspecific chronic cervicitis, and fallopian

tube without any pathology, while the tubo-ovarian nodule revealed normal ovarian stroma along with atrophic and hyalinized seminiferous tubules along with leydig cells (Figure 2). The right gonad proved to be ovotestis, confirming the diagnosis of true hermaphrodite.

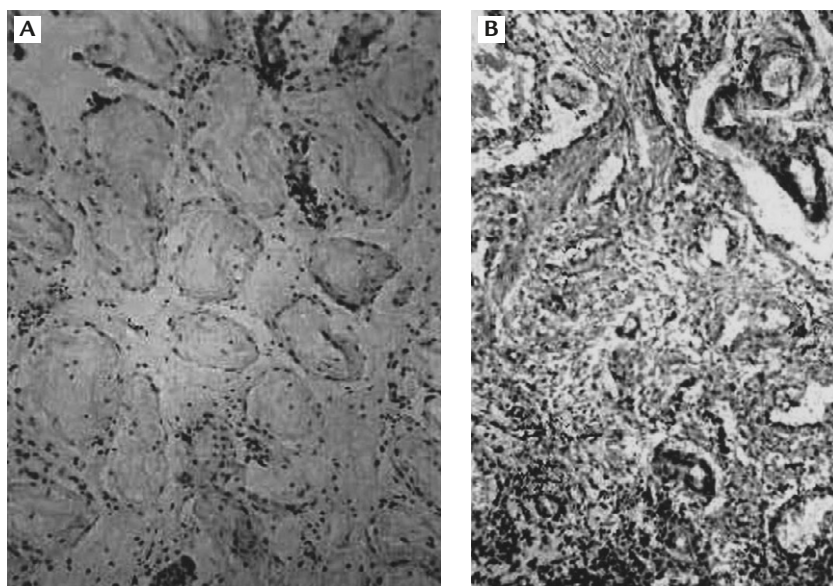
Buccal smear and cytogenetic chromosome analysis demonstrated a 46XX karyotype. Polymerase chain reaction amplification of the tumour and peripheral leucocyte deoxyribonucleic acid (DNA) with primer specific for the sex-determining region of the Y chromosome were negative.

## Discussion

True hermaphrodites are those with functional gonadal tissue of both kinds and are comparatively rare. It requires the demonstration of both well-differentiated testicular and ovarian tissue.<sup>1</sup> Few cases of tumours arising in the gonads of true hermaphrodites have been reported in the literature.<sup>2</sup> It has been suggested that paucity of reported cases of gonadal neoplasms in true hermaphrodite may be secondary to gonad destruction by expanding tumour, thereby labelling these people as “normal” or “pseudohermaphrodite”.<sup>3</sup> On account of functioning bigonads, true hermaphrodites show a wide range of morphological anomalies like a phenotypic male with ovary on one side and ovotestis on the other side.<sup>4</sup> In our patient, cryptorchidism and presence of uterus and right-sided ovotestis were confirmatory of true hermaphrodite. The cytogenetic studies confirmed 46XX karyotyping. Previous reports established the sex-determining region of the Y chromosome



**Figure 1.** Intraoperative photograph shows uterus and tubo-ovarian nodule on the right side (after excision of the lump on the left side).



**Figure 2.** (A, B) Photomicrograph of ovotestis shows normal ovarian stroma and atrophied seminiferous tubules.

(SRY gene) as the testis-determining factor.<sup>5</sup> However, it has been suggested that in most XX male individuals, the SRY gene sequence is translocated to an X chromosome.<sup>6,7</sup> This SRY gene encodes a DNA binding protein, postulated to trigger male development by repressing downstream antagonist wolffian inhibitory factor genes. Similarly, our case of male phenotype 46XX true hermaphrodite without the SRY gene is consistent with recent evidence that this gene is permissive to but not mandatory for male development. So these broad arrays of phenotypes might be explained by alteration of one or more downstream Y, X or autosomal testis determining genes. Therefore, another theory, which explained the male phenotype in our case by downstream gene on the X chromosome in which expression was influenced by X inactivation.<sup>8</sup>

The location, size and extension of tumour through the tunica in our patient indicated adjuvant therapy besides excision. It has been found that seminoma arising in cryptorchid testis tended to be relatively advanced with metastasis in a majority of patients.<sup>9</sup> In cryptorchid testis, the sites of nodal metastasis were not as predictable as in scrotal seminoma, which might complicate the selection of radiotherapy fields. As seminoma is sensitive to cisplatin-based chemotherapy and based on the reports of good prognosis for metastatic germ cell tumours of

the testis, our patient responded well to combination chemotherapy (BEP regimen) without any evidence of disease at 18-month follow-up.

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